DYNC1H1 gene

dynein cytoplasmic 1 heavy chain 1

Normal Function

The *DYNC1H1* gene provides instructions for making a protein that is part of a group (complex) of proteins called dynein. This complex is found in the fluid inside cells (cytoplasm). Dynein is turned on (activated) by attaching (binding) to another complex called dynactin. This dynein-dynactin complex binds to various materials within cells. Using energy provided by molecules called ATP, the dynein-dynactin complex moves material along a track-like system of small tubes called microtubules, similar to a conveyer belt. The dynein-dynactin complex is necessary for protein transport, positioning of cell compartments, mobility of structures within the cell, and many other cell processes. In nerve cells (neurons), dynein helps neighboring cells communicate by transporting sac-like structures called synaptic vesicles that contain chemical messengers. When synaptic vesicles are passed from one neuron to another, the dynein-dynactin complex transports the vesicle from the edge of the cell to the center where the chemical message is received.

The parts (subunits) of a dynein complex are classified by weight as heavy, intermediate, light intermediate, or light chains. Two heavy chain proteins bind together to form the core of the dynein complex. Combinations of intermediate, light intermediate, and light chains make up the rest of the complex. The protein produced from the *DYNC1H1* gene is a heavy chain. Other subunits are produced from different genes.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

spinal muscular atrophy

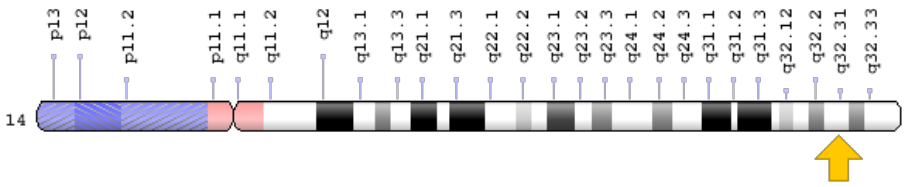
At least three mutations in the *DYNC1H1* gene have been found to cause a condition called spinal muscular atrophy, lower extremity, dominant (SMA-LED). This form of spinal muscular atrophy is characterized by leg muscle weakness that is most severe in the thigh muscles (quadriceps). The *DYNC1H1* gene mutations that cause SMA-LED replace single protein building blocks (amino acids) in the heavy chain subunit of the dynein complex. These changes disrupt the core of the dynein complex and impair its function. As a result, the movement of proteins, synaptic vesicles, and other materials within cells is reduced. A loss of synaptic vesicle transport in neurons that control muscle movement is thought to contribute to the muscle weakness

experienced by people with SMA-LED. It is unclear why this condition affects only the lower extremities.

Chromosomal Location

Cytogenetic Location: 14q32.31, which is the long (q) arm of chromosome 14 at position 32.31

Molecular Location: base pairs 101,964,528 to 102,050,798 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cytoplasmic dynein 1 heavy chain 1
- cytoplasmic dynein heavy chain 1
- DHC1
- DHC1a
- DNCH1
- Dnchc1
- DNCL
- DNECL
- DYHC
- DYHC1_HUMAN
- dynein heavy chain, cytosolic
- dynein, cytoplasmic 1, heavy chain 1
- dynein, cytoplasmic, heavy polypeptide 1
- HL-3
- p22

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Cytoplasmic Dyneins May Have Multiple Roles in the Neuron
  https://www.ncbi.nlm.nih.gov/books/NBK27955/#A1963
- Molecular Cell Biology (fourth edition, 2002): A General Model for Kinesin- and Dynein-Mediated Transport in a Typical Cell (figure)
  https://www.ncbi.nlm.nih.gov/books/NBK21710/figure/A5469/?report=objectonly
- The Cell: A Molecular Approach (second edition, 2000): Microtubule Motor Proteins (figure)
  https://www.ncbi.nlm.nih.gov/books/NBK9833/figure/A1835/
- The Cell: A Molecular Approach (second edition, 2000): Organelle Transport and Intracellular Organization
  https://www.ncbi.nlm.nih.gov/books/NBK9833/#A1836

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 2
  https://www.ncbi.nlm.nih.gov/books/NBK1285

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28DYNC1H1%5BTIAB%5D%29+OR+%28cytoplasmic+dynein+heavy+chain%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- DYNEIN, CYTOPLASMIC 1, HEAVY CHAIN 1
  http://omim.org/entry/600112

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DYNC1H1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DYNC1H1%5Bgene%5D
- HGNC Gene Family: Dyneins, cytoplasmic
  http://www.genenames.org/cgi-bin/genefamilies/set/538

- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2961
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1778
- UniProt
  http://www.uniprot.org/uniprot/Q14204

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